

Roundtable Discussion
Facilitator: Dr. Willard

DR. McCABE: Thank you very much, Dr. Cooksey.

Hunt, I'll let you take over for facilitator.

DR. WILLARD: Thank you to all three of you for framing what are not only critical issues, but ones that I suspect will generate a fair bit of interest and discussion around the table.

I'd like to start with two questions, which anyone can answer or not, that seem to me to be critical here. First of all, Joann, you argued or presented information that this has been on the agenda for anywhere between 25 years and 10 years, depending on the lag there. So the question is is there any evidence in at least the last 10 years that the education level of the workforce regarding genetics has increased or not, and has that been measured and how will we measure it over the next 10 years? Because if we're going to be serious about this, then we'll have to have a way of measuring that and assessing that.

DR. BOUGHMAN: Measurement will be a very difficult issue, but I think, starting with some of the things that you've heard this morning and in the presentations this afternoon, you will also see advances that have been made in a variety of areas, and one of our big challenges I believe is differentiating between the adage "Make it and they will come" -- build the website and people will come and learn it -- versus actually taking it to the practitioner and getting them to incorporate it in their daily practice in a competent kind of way.

As Dr. Cooksey indicated, measuring in the practice situation is going to continue to be extremely difficult. It's a hard thing to do, although there are some ways, and we'll talk a little bit about that this afternoon on ways to try and get at that and not measure it very well, but in fact at least have a baseline.

DR. WILLARD: The other question, which may take more lively discussion, and I'll start with Dr. Cooksey, is in a perfect world, how do you define success? That's directed at you, but it's really something for the committee to evaluate.

So given the amount of resources being put into education, given the obvious need for education, given what you're measuring, in a perfect world, what's the model where we would say great, that's the model we're aiming towards, because until we have a model in which we know how we'd like it to be in 2010 or 2020 or whatever the logical endpoint is, it's very hard, it seems to me, to focus either educational resources or workforce planning if we don't know what the model is that we're aiming towards.

If I have a concern about the study that you've been funded to do, and I phrase it that way because I realize some things you're supposed to be doing and other things you're not supposed to be doing, it's that it's focusing at present on genetic specialists, and yet many of the data that you gave us, some people would conclude that the battle has already been lost. If there are only 1,000 clinical geneticists in a sea of 560,000 other specialists, each of whom, to one degree or another, feels that what they work on has a significant genetics component, it may be that studying who becomes a board-certified medical geneticist or a clinical geneticist and how they spend their time and where they're located is really just scraping a little bit of the ice shaves off the top of an iceberg when in fact we should be focusing our effort on what the iceberg is.

I'm always struck and have been repeatedly by the data that you didn't mention from the AAMC, which every year, as medical students graduate from medical school and they're asked what do you want to be

when you grow up, and they all tick off a box on their chosen medical specialties, and medical genetics, over the five or six years I've tracked this number, is either 0 or 1 out of 17,000 medical students graduating every year. It's very clear.

This is at a time when we all go around, many of us, and say this is the most exciting specialty, this is a fantastic field, we quote Francis liberally, and the public is engaged and has bought into that, and yet somehow that never trickles down to the medical student who is trying to decide what he or she is going to do when they're out there in the real world.

So I'll throw that to you, and then there are hands popping up all over the place here. So I'll just have that as a point of some discussion.

DR. COOKSEY: Where to start? What I presented was only a portion of the work that we've been doing and thinking about. Let me take the last part of the question first, with the medical students.

The market really does work, and the market perception for becoming a geneticist has been modest at best. What our survey shows, and we're still analyzing it, is we asked the question would you advise a young person to go into genetics? And we actually got a little higher -- I can't remember. I think it was 75 percent said yes. We ask a lot of questions probing the esteem of the profession because we had heard anecdotally we're low paid, we don't see many patients, we're not recognized within our med school. We heard mixed pictures with geneticists.

That's not what we see in the survey. We see sort of a rather robust sense of self-esteem and a robust satisfaction except for reimbursement kinds of issues.

On the primary care situation, the AAMC, the Medical College group and many others, HRSA and others, have put strong focus that we need to build a strong primary care system to bring access to care to Americans, to help contain costs and improve quality and more holistic approaches.

What I heard last month at the International Medical Workforce Conference, which was very interesting and included a fairly large delegation of the U.S., including people from HRSA and others, in the U.S. there is real rethinking about the future of primary care providers in the country, that the concept that we have too few physicians, and particularly that we have too many physicians maybe, that was out there as a policy position and that we have too many specialists maybe, is being almost turned 180 degrees right now, that we don't have enough specialists perhaps.

Why? I don't know, but some of the contexts are the U.S. health care system, which is very different in some key ways than the Canadian or the U.K. health care system, is built on specialty care. Our reimbursement methods, the strengths of the profession, the professionalism that occurs in medical school and in residencies, is toward specialization, despite lots of efforts to get U.S. medical school graduates to go into primary care since the early '90s. The numbers went up and with the managed care and with the market looking like it was changing, with salaries for family physicians and general pediatricians going up and people going into particularly family practice, one marker profession went up and it's been going down.

It's also going down in Canada and in the U.K. despite efforts. In Canada and the U.K., they've recognized they're not producing enough homegrown medical students now and they are both increasing substantially the medical school capacity. In the U.S., the medical school capacity has been relatively stable, 15,000 or 16,000, except growth in osteopaths.

This is sort of more than you want to know, but why students aren't choosing genetics is a very important

question, but I would say that they tend to be rather smart creatures. Physicians are rather smart and they respond to what they hear and incentives that are out there.

We have heard from training programs, from genetics programs. The loss of funding has been a significant issue, funding support.

DR. WILLARD: That's a hypothesis. The alternative hypothesis is they're really excited about genetics and they've decided that the way to do that is through cardiology or hematology or medical oncology, and the answer is not necessarily to go through medical genetics.

I'll throw that out, and I've got Debra and then Ed on the list.

DR. LEONARD: So this is kind of a follow-up to that. Thinking about genetics as a specialty area isn't consistent with the future model of genetic medicine in that every physician is going to have to practice genetics, and so are workforce studies looking at the amount of genetics being practiced by all the other specialty areas? And then it comes down to an education issue, not an issue of whether there are enough specialists.

One of the issues with having it be in everybody's practice is that genetics, because the public is not well-educated about genetics, it's very time consuming because you have to start back with what's a gene and what's a mutation, not just explaining you have a mutation in such and such a gene that causes this disease.

A question. Are the current models and mechanisms for health care practice, with genetics being compartmentalized as a specialty area, and health care reimbursement issues, where you don't get paid for spending time talking to a patient, are they consistent with the anticipated future of genetic medicine?

DR. COOKSEY: Just to be clear, I fully believe that genetics will be integrated into our practice and understanding and thinking about health and medicine without question. When it will occur and how it will occur, it will occur in a jagged path probably. I have no disagreement with that.

I think when I've said that I think that it needs to be a strong specialty, is that every area that tends to be organ systems, we rely upon a cadre of specialists to help us practitioners with the more complex patients, with understanding the new and assisting with the diffusion out.

As far as, and I'm forgetting a little bit the nub of your question, is the education adequate or is the model adequate or are we studying the right things --

DR. LEONARD: Can the current health care system adapt to genetic medicine and what has to happen to make that happen?

DR. COOKSEY: We are looking. We're just starting this year to look and to talk to primary care physicians, internists, and family medicine physicians to see what they're doing now and to try to get some indication of things like what got you involved?

The data that's out there, and there is some data, says that primary care physicians become more involved with genetics when they are younger. They've had some course work in genetics. So they've been younger graduates, they're at academic medical centers, they have patients who ask them about it, and when there is something that they can do with the information.

Now, some of this research dates to the early '90s before BRCA testing and other testing was available,

but again, I think that the primary care physicians are individuals that play an extremely important role and need to be educated, but they're targeted. Their look will be targeted. There have been editorials. The primary care physician can't do him or herself a full family history. They can do targeted. So they can construct pieces of the family history.

For instance, if the family history is a core part of genetic services, I think we have to look at how that's collected within the health care system and where that data is held and how you refine it so that you don't reinvent the wheel every single time. Certainly, genetic counselors and others are very well-trained at doing the complex three-generational family history. I don't think it's necessarily efficient to expect family medicine doctors, and they've sort of said we don't have the time and we can't do it. So we have to look at this as a systems problem that we need to address and I think that efficiency is extremely important in rethinking the systems of health care.

There are some interesting models out there. The Kaiser of Northern California, Ron Bachman has a model that has a sufficient number of genetic counselors for that population. I can't remember how many covered lives he had, but there are some ways you can look at some good models and sort of get the numbers that might be needed and see how they're used when you remove reimbursement constraints and whatever and when you have a defined population group and a care system that has responsibility and it takes responsibility for managing the patients.

So I think there are some models that are out there, but I think it requires new thinking, less focus on the silos of who does what, more recognition that the incentives will be for subspecialists and specialist physicians to take it up. They will have incentives to do that. For the primary care physicians, there are different incentives and different issues, and you've got to recognize the incentives and go with those, whether those are professional or financial or whatever. That's what drives people.

DR. WILLARD: Thank you.

Ed?

DR. McCABE: Two things. One is that I firmly believe we've got to improve genetics education. Everyone has to be knowledgeable in genetics.

I'd also like to think there's going to be a role for the geneticist in the future, and the analogy is the radiologist. We all look at x-rays, but when we really want to know what's in that x-ray, we go to the radiologist, and I think that's the way we're going to dig in genetic information.

Having said that, though, I think there's a cultural problem in genetics. Medical students go to medical school because they want to make people feel better. Geneticists are diagnosticians. Neurology went through this a few years ago when they were losing all of the -- they couldn't do anything either, and they developed an area of interventional neurology, because otherwise that was going to be lost to the neurosurgeons and the others.

I think if we continue to focus on diagnostics in genetics, we are doomed to failure because it will be a very small part of the physician graduates who want to go out and never make anybody feel any better, and that's a problem. Outside of biochemical genetics, it was mentioned today, what we do is manage. We don't really make people feel better.

DR. WILLARD: Thank you.

I've have Reed, Francis, Barbara, Kimberly, Chris, and Brad in rapid order. Reed?

DR. TUCKSON: Rapidly, how would you assess, any of you, the adequacy of the genetic-based education in health professional schools today? Are you convinced that the leaders of medical schools and nursing schools have understood the inevitability that medicine is a genetic-based discipline now and that if you're going to learn heart disease, there is a genetic basis of heart disease? If you're going to learn cancer, there's a genetic basis of cancer. If you're going to learn liver disease -- there's a genetic basis of all these things. Has there been a revolution or are we still at the point where today's graduates, we will still be having to plan on continuing medical education catch-up, even for this class, or have they accepted this?

DR. BOUGHMAN: I will give you a little bit of data this afternoon, but the current students, at least in medical school, by the counts that we have available, are not only receiving in nearly every medical school the basics of genetics, they in fact are being tested on at least aspects of the basics of genetics, even in the national medical examinations by the national boards. That is a great improvement over five years ago.

The real trick, though, however, is not back to the definition of the gene or the basics in didactic lecture format. It is actually during the training and the practitioner aspects of this. How do I incorporate this? That immediately becomes both a curricular issue in the training program and a process that the professional guidelines that are out there that help determine what the services that a patient with Disease X should be able to receive in order to be cared for adequately.

DR. TUCKSON: Nursing schools?

DR. COOKSEY: I can comment on nursing. Nursing has studied this issue and their consensus is that general nursing education at the baccalaureate level or the associate degree level has had very limited inclusion of genetics in the curriculum, and they are trying to address that.

I think another issue that should be considered when we think about educational preparedness and competency for the workforce is to recognize that you have 2.5 million or so practicing nurses out there, and you need some level of awareness for all professionals within various areas.

But something else that has to be right up there as well is recognizing that within professions there needs to be again this cadre of genetics specialists, I think both to help educate and to help move the profession along and set the vision and the pace. So whether it's pharmacists, nurses, other professionals, it's that higher level, whether it's master or at least trained, who are doing clinical practice that's in more sophisticated genetics to help do the training and education and keep the profession aware of the changes that are happening. So it's not just the body of everyone, like all med students. It's that specialty training.

DR. TUCKSON: I can't ask any more questions, but what I hear, though, and what I don't hear from these answers is I think I hear that it's coming. What I don't hear is we're on top of it.

DR. SHEKAR: Well, let me respond to that from our perspective that both the projects that I mentioned, the Genetics in Primary Care as well as the GIFT program, succeeded and are succeeding because of the commitment of the university administration over and above the faculty that are applying for those projects. So those are applied for by those universities, and the only way that those programs are getting those is obviously the quality of the applications, but also the support from people from within to provide that support to those projects. So it's very clear, Reed, that in order for genetics to move forward through those venues, there obviously needs to be support over and above the faculty investigative level.

DR. BOUGHMAN: The NCHPEG talk this afternoon will help answer that.

DR. WILLARD: I want to keep this going because my watch says we have 10 minutes.

Francis Collins?

DR. COLLINS: I'll try to be quick. I think, Hunt, you asked the right question at the beginning of this discussion about what is the model that we would like to see happen in the course of the next decade or so, as opposed to what path are we on right now, because I think the path we're on right now doesn't look very encouraging. The statistics quoted about medical students running away as fast as they can from the discipline of medical genetics are sobering indeed.

I think a major reason for that is the absence of effective role models in their vicinity. As medical students, we're all influenced by the people we see in our environment who sort of are the kinds of docs that we would like to become, and sadly, I think medical geneticists, most of them pediatricians, are seen as taking care of relatively rare conditions. They're seen, sadly, as practicing the genetics of yesterday, instead of the genetics of tomorrow. The genetics of tomorrow seems to have been grabbed up by other disciplines, BRCA1 and oncology as the most obvious example, but there are others. I think students are pretty clever to figure out sort of which way the wind is blowing.

So I think the model that you ask for, which most people I think would now endorse, is the sort of radiology example, where everybody who is a primary care provider, and that's not just M.D.s, need to acquire genetic skills, and when Joe McNerney talks about NCHPEG, I'm sure he'll go through some of the ways in which that organization is trying to push that agenda forward, but that there will have to be experts, specialists, who are in a position to sort out the complex circumstances that are going to happen very frequently.

I see two major problems with that model. One is reimbursement, and that would be a very appropriate topic for this committee to think about in terms of how are we going to take something which is time-intensive and procedure-limited and turn that into something that could be adequately reimbursed as an incentive both for the primary care provider and the expert to actually be compensated for what they do.

The other barrier, though, that I see is what appears to be at the moment a fairly strong level of resistance on the part of those who would become the experts to taking on that new role, to giving up the role of being the academic clinical geneticist taking care of the rare disease and becoming really, in a much more different kind of model, the place to which a wide variety of complex conditions, most of them probably affecting adults, would get referred for sorting out. Also, as part of that, abandoning, sadly, the model where you can't see a patient in less than an hour and a half, which has to be abandoned if this whole thing is going to work.

So the question I have after my little speech here is to ask Judith, in your survey of medical geneticists, do you in any way get feedback from them about their receptivity to that kind of a very different kind of professional role in the future?

DR. COOKSEY: We did not probe those questions on the survey. I think the survey methodology would be very limited to that, but it could have been something -- I'd have to go back and look at the survey and see if we can get some hints. We certainly asked them attitudes, beliefs, their sense of the quality of care that's provided, the quality of genetics care that's provided. I'd have to think if we covered some of those others and I'd be happy to do it.

I think that one example to think about in this issue might be the infectious disease doctors and how they stepped up to the plate with HIV and how they're continuing to step up to the plate with emerging

infections, with bioterrorism, with other things. You had a very relatively small specialty that, as another example to radiology, treats all ages, everybody can get infectious diseases, and they can be mild or they can be serious and life-threatening. They're treatable.

They stepped up to the plate and they helped train others, which geneticists are doing. There's a continuing need for infectious disease experts because of some challenges that we're facing, but leadership from that community, at a time that many physicians and other health professionals did not want to see HIV and AIDS patients because of personal risk or perceived personal risk, they came up. That was important and we've diffused the treatment of HIV out into fairly broad medical practice now with the specialty.

Francis, the question that I pose to you, I think it's a little premature to answer Hunt's question about what's the right model. We can think more about that. We're keenly aware that the current model isn't the best model. I happen to think there's value in studying it.

You haven't addressed the issue of the large group of specialist physicians who are taking on genetics and how geneticists really have an opportunity to work with that group because that's where things are going right now.

DR. WILLARD: Great. Thank you.

Barbara?

MS. HARRISON: One of my main concerns, as well as we've been talking a little bit about increasing the number of medical geneticists, but also increasing the number of genetic counselors, and I think the root of that problem is reimbursement, which we may talk about more later. I don't want to spend the time now.

But I one of the highlights when you were talking was this Bureau of Labor Statistics and how we are just not on there and how that's used by a lot of career counselors and guidance counselors, which, especially from the genetic counseling standpoint, are who we need to reach. I was just wondering if you had any ideas about how we can get on that list or kind of what the issue is there.

DR. COOKSEY: The Bureau of Labor Statistics is down the street a bit. The problem is that the data they collect, they fund every state, usually an Office of Employment Security, to do surveys of employers as to what categories of employees they employ by occupation, and then they fill in with Census data the self-employed.

We could certainly talk with them. I think the numbers are probably too low, and when they look at physicians and when they look at the 600,000 physicians or nearly 600,000, they look at physicians, they look at pharmacists, they look at dentists. They don't look at the subspecialty level. So it's a very coarse measure that looks at every occupation in the country, including some small-level ones. You know, hair stylists, manufacturers of stainless steel. I mean, this is labor statistics information for our whole economy.

So we could, but I don't think that's quite the way to go to get on their radar. I just think that's not quite the -- it won't succeed, I think.

MS. HARRISON: Okay. Just real quick, would allied health professionals be on that list?

DR. COOKSEY: Specific allied health professions are. They have diagnostic professional groups and

they have technicians and technologists and sort of more support level. Again, it needs to be a fairly large group, but there's about 30 different professional groups that they do collect data on, and I can share that and see if it looks like it makes sense.

DR. WILLARD: Thank you.

Kimberly?

MS. ZELLMER: I just had kind of a quick comment. I think that from the lay person's point of view, having been through amniocentesis and all that with my kids, it seems like there are certain areas that maybe there's a better job of getting genetic information out there and doing a much better job.

But I think in the case of rare diseases, I think that they are still way far behind, and I think that the primary care physicians, I think they're probably good to refer patients on to specialists, but I think even in rare diseases, the specialists aren't making the diagnosis of rare diseases. In our case, we went to three different pediatric neurologists at teaching hospitals before we got a diagnosis of a rare genetic disorder.

Then the other thing that I think is important for a primary care physician's education is not only knowing to refer patients and having that basic genetic information, but part of the problem that I know that we've had and also I know from other families with kids with genetic disorders is once you've been diagnosed with a genetic disorder, many primary care physicians don't know what to do with you.

We've been to three different pediatricians to find one who's comfortable giving us advice on whether or not our daughter should get immunizations or not or what types of medications it's okay for her to take, and the pediatricians like to defer to the pediatric neurologists, but the pediatric neurologists know nothing about more common medications or they know very little about immunizations. So it's hard to find someone who's going to make those basic decisions.

So I think you've got issues of not only finding people to recognize less common genetic disorders, but I think also teaching primary care physicians what their role is going to be once they're dealing with a patient who has a genetic disorder.

DR. McCABE: Thank you very much.

With that comment, I think we're going to wrap up. Thank you, Hunt, and thank you to our panelists. Dr. Boughman, we'll see you again after lunch.

We only have 45 minutes for lunch today. So that's going to be a challenge to everyone. We do need to be back here sharply at 1 o'clock, though, because that's when we have our public comment session. We have seven people in a 30-minute session this afternoon, so I would ask all of our public commentators to please try over lunch to think about how you can keep your comments to two to two and a half minutes so there is time for us to have discussion.

Thank you.

(Whereupon, at 12:15, the meeting was recessed for lunch, to reconvene at 1:00 p.m.)